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CASE REPORT

Association head and neck angiosarcoma and nevoid basal cell carcinoma syndrome (Gorlin syndrome)

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Summary We describe herein an association of a nevoid basal cell carcinoma syndrome and an angiosarcoma arising in muscle of neck. This inextirpable tumour showed a rapid progression. The patient died 3 months after diagnosis despite two lines of chemotherapy. This association was not previously described.

Introduction

Soft tissue sarcomas are rare (estimated incidence 3-4.5/100 000).[1] Five pour-cents of all soft tissue sarcomas arise in the head and neck region.[1–3] Little is known about aetiology, prognostic factors and treatment results of those cancers.[1–3]

About 3% of all soft tissue sarcomas are associated with genetic disease, essentially Recklinghausen disease. Others genetic diseases are exceptionally associated with soft tissue sarcomas: polypadenatous coli, Werner syndrome, bilateral retinoblastoma...[1]

We describe herein an association of head and neck angiosarcoma and nevoid basal cell carcinoma syndrome (NBCC syndrome).

Case report

A 41-year-old man was refereed for management of a large tumour of neck muscles (Fig. 1). The tumour had appeared 3 months before on the left side of the neck, was painless, and showed a rapid initial

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Figure 1 Eighteen FDG-positron emission tomography: initial locoregional extension of the angiosarcoma.

growth. A biopsy was performed and the histological examination revealed an epithelioid angiosarcoma. Further evaluation with head and neck CT scan and positron emission tomography revealed an 8 cm necrotic tumour with multiple lymph nodes extension (supra-clavicular and upper mediastinum) and large involvement of neck muscles.

This patient presented with previous history of sporadic NBCC syndrome, with macrocephaly due to Arnold—Chiari malformation, multiple keratocysts of the jaw and 12 previous basal cell carcinomas of face and chest treated by surgery.

The angiosarcoma was considered as inoperable. Thus, the patient received two cycles of polychemotherapy regimen including adriamycin, ifosfamid and dacarbazine. Despite the chemotherapy, we observed a rapid growth, with apparition of controlateral lymph node extension, involvement into upper mediastinum and liver metastases. A second-line chemotherapy with paclitaxel failed to stop the progression and the patient died 3 months after diagnosis.

Discussion

The nevoid basal cell carcinoma syndrome (NBCC), also known as Gorlin syndrome and the basal cell nevus syndrome, is an autosomal dominant disorder that predisposes to basal cell carcinoma (BCCs) of the skin, medullobastomas, and ovarian fibromas.[4] Its prevalence has been estimated at 1 per 56 000. One to 2% of medulloblastomas and 0.5% of basal cell carcinomas are attributable to the syndrome.[4—6]

BCCs in this disorder are multiple, occur relatively early compared with sporadic tumours of the same type, and tend to develop after exposure to ionizing radiation with a very brief latency period.[4—7]

In addition to benign and malignant tumours, malformations are a striking component. The syndrome is associated with pits of the palms and soles, keratocysts of the jaw and other dental malformations, cleft palate, characteristics coarse facies, strabismus, dysgenesis of corpsus callosum, calcification of the falx cerebri, spina bifida occulta and other spine anomalies, bifid ribs, ectopic calcifications, mesenteric cysts, macrocephaly and generalized overgrowth.[4—7]

The extent of expression of the many features of the syndrome is variable, but the severity tends to breed true within families. There are occasional sporadic cases. The behaviour of tumours in NBCC suggests that the underlying defect in this disorder may be a mutation in a tumour suppressor gene. Mapping of the NBCC gene to chromosome 9 and the demonstration of the exact same region is deleted in a high percentage of BCCs and other tumours related to the disorder provided strong evidence of this gene may be a necessary if not sufficient event for the development of BCCs.[8]

The diagnosis of NBCC should be considered in anyone below the age of 30 with a single BCC and in older individuals with multiple BCCs. Medulloblastomas, keratocysts of the jaw and typical skeletal anomalies should raise the suspicion of NBCC even in absence of BCCs. Palmar and plantar pits are pathognomonic.

The most important follow-up study in affected individuals is dermatologic examination for BCCs at intervals of 6 months to 1 year.[7]

Head and neck soft tissue sarcomas are rare.[1—3] Their treatment required a large resection in a multimodal approach. Actually, theirs pseudocapsules represent an artificial and inadequate limit. Those cancers spread along facial planes, muscles, nerves and vessels, and reduce the possibility of resection with adequate margins. So, the prognosis of head and neck soft tissue sarcoma is poor.[2,3]

Two histological subtypes are particularly frequent in head and neck: rhabdomyosarcoma and angiosarcoma.[2,3] Multicentric angiosarcoma occurs in the scalp and the face of elderly men, where unrelenting progression can cause severe ulceration and infection. Surgical treatment is not

possible in most cases as a result palliative radiotherapy and chemotherapy are indicated.[9,10]

Angiosarcomas represent 1% of all soft tissue sarcomas. Their clinical outcomes are poor due to a rapid growth, with high risk of metastatic extension. The median overall survival of all clinical types of angiosarcomas is ranged between 15 and 30 months.[11]

Adriamycin and ifosfamid-based regimen achieve a response rate about 30% for metastatic or locally advanced soft tissue sarcoma.[1] Paclitaxel seems to be effective for treatment of scalp angiosarcoma.[10]

Our case report show the poor outcome of locally advanced head and neck soft tissue sarcoma, especially head and neck angiosarcoma. Association NBCC syndrome and angiosarcoma was not previously described. In NBCC, other soft tissue tumours probably occur to excess include fibrosarcomas, rhabdomyosarcomas, and cardiac fibroma. To conclude, in patient with previous history of Gorlin syndrome, the apparition of a soft tissue tumour requires absolutely a biopsy in order to treat early a soft tissue sarcoma.

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